

GENETIC TESTING PRACTICE ACTIVITIES
DIVISION OF PUBLIC HEALTH PARTNERSHIPS-LABORATORY SYSTEMS
NATIONAL CENTER FOR HEALTH MARKETING

- Who we are** The genetics group within the National Center for Health Marketing’s Division of Public Health Partnerships includes two board-certified laboratory geneticists and three ORISE fellows with expertise in genetics, genetic counseling, nursing, and molecular technology.
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- What we do** We engage the public and private sector to promote the quality of genetic testing in clinical and public health practice. We accomplish this by conducting activities that
- monitor and evaluate current and emerging laboratory practices associated with genetic testing
 - assure appropriate ordering, reporting, and use of genetic tests and results
 - assure availability and access to a variety of quality control (QC) materials
 - assure availability and access to quality genetic testing for rare diseases
 - assure a competent workforce
 - develop and evaluate voluntary and regulatory standards, guidelines, and policies
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- Our partners and customers** Much of our work includes ongoing collaborations with a diversity of partners that include
- State and local public health professionals and laboratories
 - Clinical laboratory services
 - Providers of genetic testing services
 - Users of genetic testing services
 - Policy makers that address genetic testing issues
 - International public health organizations
- Monitoring and evaluation activities** We are working with external partners to prepare reports on the current and emerging state of laboratory practices relevant to genetic testing. These efforts inform program development and evaluation for CDC, other federal and State agencies, the private sector, and international efforts.

Major Accomplishment, 2004:

Co-authored “Ensuring the Quality of Genetic Testing in the United States,” and “Carrier Testing for Cystic Fibrosis: Transition from Research to Clinical Practice,” chapters in the *CDC Report on Genomics and Public Health 2003*, published March 2004.

**Ordering,
reporting and
use of genetic
tests**

We are facilitating the development of laboratory and health provider networks to address public health concerns in assuring appropriate ordering, reporting, and use of genetic tests and results. We are also working with the international community to address similar issues pertaining to cross-border flow of genetic test referrals.

Major Accomplishments, 2004:

- As a result of a May 2003 national conference held in collaboration with Mt. Sinai's College of Medicine, efforts are underway to create more effective requisitions for genetic testing services and improve test reports by assuring critical test result information is clearly and concisely presented.
 - We partnered with the European Union cystic fibrosis network to perform a comparative analysis of U.S. and European reporting practices. Results from this study will assist policy makers in determining where international consensus may be achieved.
 - We accepted a co-leadership position in addressing international test result reporting issues as part of our involvement with the Organization for Economic Cooperation and Development's (OECD) Steering Group on Quality Assurance and Proficiency Testing in Molecular Genetic Testing.
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QC materials

We are spearheading a community process to implement a sustainable mechanism to provide appropriate quality control materials and guidelines for use in routine genetic testing, inter-laboratory comparisons of quality assessment, and test development.

Major Accomplishments, 2004:

- Together with key members of the genomic/molecular diagnostic community we hosted two conferences entitled "QC Materials for Genetic Testing." The conferences were held to develop recommendations relative to current and future needs for genetic testing QC materials and establish a sustainable, practical process to make QC materials available to genetic testing laboratories.
 - Based on the recommendations from the above conferences, a framework has been established to collect, store, verify, and distribute certain cell lines that may be used for genetic quality control and assessment procedures. A CDC-based Coordination Program will facilitate the collection and verification of and access to the materials; private sector repositories will store and distribute them.
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Rare disease genetic testing We are addressing an urgent need to assure and improve availability of quality genetic testing for rare diseases.

Major Accomplishment, 2004:

In collaboration with other HHS agencies, CDC CIOs, professional organizations, patient advocacy groups, and Emory University, we convened a conference entitled "Promoting Quality Laboratory Testing for Rare Diseases: Keys to Ensuring Quality Genetic Testing." The conference brought together a broad scope of stakeholders to address crucial issues surrounding the availability of and access to quality genetic testing for rare diseases, which affect 25 million (or 1 in every 12) people in the United States.

Workforce competencies Working with our external partners, educational programs are developed, taught, and evaluated to promote workforce competency of users and providers of genetic testing services.

Major Accomplishments, 2004:

- With assistance from the American College of Medical Genetics and the American Medical Association, we have distributed over 4,000 copies (1,415 copies in 2004) of our award-winning interactive CD-ROM entitled "Genetics in Clinical Laboratory Practice: A Team Approach." This interactive multimedia educational tool, developed through a cooperative agreement with Dartmouth Medical School, emphasizes the uniqueness of the genetic testing process and increases the knowledge of healthcare providers using genetic tests in clinical practice.
 - Since September 2002, a webcast produced by the Division of Laboratory Systems entitled "A New Era in Newborn Screening - Saving Lives, Improving Outcomes," has been viewed by a total of 690 health professionals (249 in 2004). The satellite program, also available on CD ROM, provides information to improve recognition, detection, and diagnosis leading to early intervention and effective management of metabolic disease, endocrine disorders, and hemoglobinopathies in newborns.
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**Standards,
Guidelines,
and Policies**

We are working with federal and State agencies, professional organizations and international groups to develop voluntary and regulatory standards, guidelines, and policies to improve the quality of laboratory practice.

Major Accomplishments, 2004:

- We continued to provide the Centers for Medicare & Medicaid Services technical consultation on the development of quality and personnel requirements for genetic testing under the Clinical Laboratory Improvement Amendments of 1988 (CLIA).
 - We participated as U.S. representatives to the OCED's Steering Group's meeting on Quality Assurance and Proficiency Testing for Molecular Genetic Testing.
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**Future
activities**

In 2005 we plan to

- Conduct health services research through a cooperative agreement with the Wadsworth Center, New York State Department of Health, entitled "Improving the Quality of Genetic Testing and Assuring its Appropriate Integration into Clinical and Public Health Practice"
 - Convene a conference entitled "Best Practices in Ordering and Reporting Genetic Tests and Results"
 - Publicly launch the QC Materials Coordination website, which will facilitate information exchange and communication about QC materials needs and availability
 - Disseminate validated cell lines for genetic testing quality control and quality assessment activities
 - Facilitate development of public/private processes to improve genetic testing for rare diseases
 - Collaborate with public health laboratories to identify the resources and training needs to ensure continued capacity to support public health services
 - Continue to collaborate with other federal agencies, professional organizations, and users and providers of laboratory services to assure quality genetic testing services and protect patient safety
 - Continue to collaborate with OECD, Eurogentest, ISO and other international groups to harmonize and promote quality and good practices of genetic testing services worldwide
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**For more
information**

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